



SEQUENCE LISTING

<110> Gravel, Roy A.
Rozen, Rima
LeClerc, Daniel
Goyette, Philippe
Campeau, Eric

<120> HUMAN METHIONINE SYNTHASE: CLONING, AND
METHODS FOR EVALUATING RISK OF NEURAL TUBE DEFECTS,
CARDIOVASCULAR DISEASE, AND CANCER

<130> 50004/002005

<140> 10/607,712

<141> 2003-06-27

<150> 08/980,326

<151> 1997-11-26

<150> 60/031,964

<151> 1996-11-27

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<223> Wild type methionine synthase polypeptide.

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 Tyr Leu Pro Leu Ser Gln Ala Arg Lys Ser Gly Phe Gln Met Asp Trp
 930 935 940
 Leu Ser Glu Pro His Pro Val Lys Pro Thr Phe Ile Gly Thr Gln Val
 945 950 955 960
 Phe Glu Asp Tyr Asp Leu Gln Lys Leu Val Asp Tyr Ile Asp Trp Lys
 965 970 975
 Pro Phe Phe Asp Val Trp Gln Leu Arg Gly Lys Tyr Pro Asn Arg Gly
 980 985 990
 Phe Pro Lys Ile Phe Asn Asp Lys Thr Val Gly Gly Glu Ala Arg Lys
 995 1000 1005
 Val Tyr Asp Asp Ala His Asn Met Leu Asn Thr Leu Ile Ser Gln Lys
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 Tyr Ser Ser Ile Met Val Lys Ala Leu Gly Asp Arg Leu Ala Glu Ala
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26

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 35 40 45
 Asp Ile
 50

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 <212> PRT
 <213> Mus musculus

<400> 68
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 <212> PRT
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 <213> Mycobacterium leprae

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 <213> Caenorhabditis elegans

<400> 71
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 <211> 50
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 <213> Hemophilus influenzae

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 Xaa at position 920 is either His or Asp.

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Glu	His	Phe	Arg	Gly	Gln	Glu	Phe	Lys	Asp	His	Ala	Arg	Pro	Leu	Lys		
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Gly	Asn	Asn	Asp	Ile	Leu	Ser	Ile	Thr	Gln	Pro	Asp	Val	Ile	Tyr	Gln		
65					70					75					80		
Ile	His	Lys	Glu	Tyr	Leu	Leu	Ala	Gly	Ala	Asp	Ile	Ile	Glu	Thr	Asn		
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Thr	Phe	Ser	Ser	Thr	Ser	Ile	Ala	Gln	Ala	Asp	Tyr	Gly	Leu	Glu	His		
			100					105					110				
Leu	Ala	Tyr	Arg	Met	Asn	Met	Cys	Ser	Ala	Gly	Val	Ala	Arg	Lys	Ala		
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Ala	Glu	Glu	Val	Thr	Leu	Gln	Thr	Gly	Ile	Lys	Arg	Phe	Val	Ala	Gly		
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Ala	Leu	Gly	Pro	Thr	Asn	Lys	Thr	Leu	Ser	Val	Ser	Pro	Ser	Val	Glu		
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Arg	Pro	Asp	Tyr	Arg	Asn	Ile	Thr	Phe	Asp	Glu	Leu	Val	Glu	Ala	Tyr		
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Gln	Glu	Gln	Ala	Lys	Gly	Leu	Leu	Asp	Gly	Gly	Val	Asp	Ile	Leu	Leu		
			180					185					190				
Ile	Glu	Thr	Ile	Phe	Asp	Thr	Ala	Asn	Ala	Lys	Ala	Ala	Leu	Phe	Ala		
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Leu	Gln	Asn	Leu	Phe	Glu	Glu	Lys	Tyr	Ala	Pro	Arg	Pro	Ile	Phe	Ile		
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Gly	Glu	Gly	Phe	Val	Ile	Ser	Val	Ser	His	Gly	Glu	Pro	Leu	Cys	Ile		
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Gly	Leu	Asn	Cys	Ala	Leu	Gly	Ala	Ala	Glu	Met	Arg	Pro	Phe	Ile	Glu		
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Ile	Ile	Gly	Lys	Cys	Thr	Thr	Ala	Tyr	Val	Leu	Cys	Tyr	Pro	Asn	Ala		
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Ala	Lys	His	Leu	Lys	Asp	Phe	Ala	Met	Asp	Gly	Leu	Val	Asn	Ile	Val		
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Ala	Val	Lys	Asn	Cys	Lys	Pro	Arg	Val	Pro	Pro	Ala	Thr	Ala	Phe	Glu		
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Thr	Asn	Phe	Val	Asn	Ile	Gly	Glu	Arg	Cys	Asn	Val	Ala	Gly	Ser	Arg		
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Lys	Phe	Ala	Lys	Leu	Ile	Met	Ala	Gly	Asn	Tyr	Glu	Glu	Ala	Leu	Cys		
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Val	Ala	Lys	Val	Gln	Val	Glu	Met	Gly	Ala	Gln	Val	Leu	Asp	Val	Asn		
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Met	Asp	Asp	Gly	Met	Leu	Asp	Gly	Pro	Ser	Ala	Met	Thr	Arg	Phe	Cys		
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Asp	Ser	Ser	Asn	Phe	Ala	Val	Ile	Glu	Ala	Gly	Leu	Lys	Cys	Cys	Gln		
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Glu	Ala	Thr	Glu	Lys	Leu	Leu	Arg	Tyr	Ala	Gln	Thr	Gln	Gly	Thr	Gly	
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Gly	Lys	Lys	Val	Ile	Gln	Thr	Asp	Glu	Trp	Arg	Asn	Gly	Pro	Val	Glu	
			660					665					670			
Glu	Arg	Leu	Glu	Tyr	Ala	Leu	Val	Lys	Gly	Ile	Glu	Lys	His	Ile	Ile	
		675					680					685				
Glu	Asp	Thr	Glu	Glu	Ala	Arg	Leu	Asn	Gln	Lys	Lys	Tyr	Pro	Arg	Pro	
	690					695					700					
Leu	Asn	Ile	Ile	Glu	Gly	Pro	Leu	Met	Asn	Gly	Met	Lys	Ile	Val	Gly	
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Asp	Leu	Phe	Gly	Ala	Gly	Lys	Met	Phe	Leu	Pro	Gln	Val	Ile	Lys	Ser	
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Ala	Arg	Val	Met	Lys	Lys	Ala	Val	Gly	His	Leu	Ile	Pro	Phe	Met	Glu	
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Lys	Glu	Arg	Glu	Glu	Thr	Arg	Val	Leu	Asn	Gly	Thr	Val	Glu	Glu	Glu	
		755					760					765				
Asp	Pro	Tyr	Gln	Gly	Thr	Ile	Val	Leu	Ala	Thr	Val	Lys	Gly	Asp	Val	
	770				775						780					
His	Asp	Ile	Gly	Lys	Asn	Ile	Val	Gly	Val	Val	Leu	Gly	Cys	Asn	Asn	
785					790					795					800	
Phe	Arg	Val	Ile	Asp	Leu	Gly	Val	Met	Thr	Pro	Cys	Asp	Lys	Ile	Leu	
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Ile	Thr	Pro	Ser	Leu	Asp	Glu	Met	Ile	Phe	Val	Ala	Lys	Glu	Met	Glu	
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865					870				875						880	
Xaa	His	Val	Leu	Asp	Ala	Ser	Lys	Ser	Val	Val	Val	Cys	Ser	Gln	Leu	
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Tyr	Glu	Asp	Ile	Arg	Gln	Xaa	Xaa	Tyr	Glu	Ser	Leu	Lys	Glu	Arg	Arg	
		915				920						925				
Tyr	Leu	Pro	Leu	Ser	Gln	Ala	Arg	Lys	Ser	Gly	Phe	Gln	Met	Asp	Trp	
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 Phe Pro Lys Ile Phe Asn Asp Lys Thr Val Gly Gly Glu Ala Arg Lys
 995 1000 1005
 Val Tyr Asp Asp Ala His Asn Met Leu Asn Thr Leu Ile Ser Gln Lys
 1010 1015 1020
 Lys Leu Arg Ala Arg Gly Val Val Gly Phe Trp Pro Ala Gln Ser Ile
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 Gln Asp Asp Ile His Leu Tyr Ala Glu Ala Ala Val Pro Gln Ala Ala
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 Glu Pro Ile Ala Thr Phe Tyr Gly Leu Arg Gln Gln Ala Glu Lys Asp
 1060 1065 1070
 Ser Ala Ser Thr Glu Pro Tyr Tyr Cys Leu Ser Asp Phe Ile Ala Pro
 1075 1080 1085
 Leu His Ser Gly Ile Arg Asp Tyr Leu Gly Leu Phe Ala Val Ala Cys
 1090 1095 1100
 Phe Gly Val Glu Glu Leu Ser Lys Ala Tyr Glu Asp Asp Gly Asp Asp
 1105 1110 1115 1120
 Tyr Ser Ser Ile Met Val Lys Ala Leu Gly Asp Arg Leu Ala Glu Ala
 1125 1130 1135
 Phe Ala Glu Glu Leu His Glu Arg Val Arg Arg Glu Leu Trp Ala Tyr
 1140 1145 1150
 Cys Gly Ser Glu Gln Leu Asp Val Ala Asp Leu Arg Arg Leu Arg Tyr
 1155 1160 1165
 Lys Gly Ile Arg Pro Ala Pro Gly Tyr Pro Ser Gln Pro Asp His Thr
 1170 1175 1180
 Glu Lys Leu Thr Met Trp Arg Leu Ala Asp Ile Glu Gln Ser Thr Gly
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 1205 1210 1215
 Gly Leu Tyr Phe Ser Asn Leu Lys Ser Lys Tyr Phe Ala Val Gly Lys
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 Asp
 1265

<210> 75
 <211> 3856
 <212> DNA
 <213> Homo sapiens

<220>
 <221> variation
 <222> (1)...(3856)
 <223> nnn at positions 2640-2642 is either AAT or no
 nucleotides; n at position 2756 is either A or G;
 n at position 2758 is either C or G.

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 atccagcggg agaagctaaa cgaagaacac ttccgaggct aggaatttaa agatcatgcc 180
 aggccgctga aaggcaacaa tgacatttta agtataactc agcctgatgt catttaccaa 240
 atccataagg aatacttgct ggctggggca gatatcattg aaacaaatac ttttagcagc 300

actagtattg	cccaagctga	ctatggcctt	gaacacttgg	cctaccggat	gaacatgtgc	360
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aggccggatt	ataggaacat	cacatttgat	gagcttggtg	aagcatacca	agagcaggcc	540
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tatgcattga	ggaagaacat	atctgtggct	gaggttgaga	aatggcttgg	acccattttg	3780
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3856

<210> 76

<211> 10

<212> DNA

<213> Homo sapiens

<400> 76

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10